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Ichthyosis-short stature-brachydactyly-microspherophakia syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Ichthyosis-short stature-brachydactyly-microspherophakia syndrome. ORPHA:363992*

A rare, syndromic ichthyosis characterized by a collodion membrane at birth, generalized congenital ichthyosis, microspherophakia, myopia, ectopia lentis, short stature with brachydactyly and joint stiffness, and occasionally mitral valve dysplasia.